Complete and print.

Reset Form

MAYO CLINIC LABORATORIES

Connective Tissue/Cerebrovascular Disease Genetic Testing Patient Information

Instructions: This form is intended to be completed by the ordering healthcare professional. Accurate interpretation and reporting of genetic results is contingent upon ancestry, the reason for testing, clinical information, and biological family history. To help provide the best possible service, supply the information requested below and **send paperwork with the specimen, or return by fax to Mayo Clinic Laboratories, Attn: Molecular Technologies Laboratory Genetic Counselors at 507-284-1759. Phone: 800-533-1710 / International clients: 855-379-3115 or +1-507-284-9273, or email mliintl@mayo.edu.**

Patient Information

Patient Name (Last, First Middle)		Birth Date (mm-dd-yyyy)
Preferred Name	Medical Record Number (if Birth Date is not available)	
Sex Assigned at Birth ☐ Male ☐ Female ☐ Choose not to disclose ☐ Other, specify:	Legal/Administrative Sex ☐ Male ☐ Female ☐ Nonbinary ☐ Choose not to disclose ☐ Other, specify:	
Gender Identity (optional)	Pronouns (optional)	

Referring Healthcare Professional Information

Referring Healthcare Professional Name (Last, First)	Phone	Fax*
Genetic Counselor/Other Healthcare Professional Name (Last, First)	Phone	Fax*

*Fax number given must be from a fax machine that complies with applicable HIPAA regulations.

Reason for Testing Check all that apply.

☐ Diagnosis ☐ Biological family history** ☐ Sudden death

**Genetic testing should be performed on an affected biological family member first, when possible. FMTT / Familial Variant, Targeted Testing should be ordered when there is a previous positive genetic test result in the family.

Clinical History

Diagnosis/Suspected Diagnosis

☐ Marfan Syndrome ☐ Loeys-Dietz Syndrome ☐ Osteogenesis Imperfecta
☐ Ehlers-Danlos Syndrome ☐ Familial thoracic aortic aneurysm and dissection ☐ Cerebrovascular disease/stroke
☐ Other:

Indicate whether the following are present:

☐ Aortic diameter at sinuses of Valsalva Z-score ≥ 2
☐ Aortic dissection
☐ Ectopia lentis
☐ Systemic score ≥ 7 points (see table on page 2 for calculation)
☐ Aortic dilatation/aneurysm (Z-score < 2)
☐ Biological family history of independently diagnosed Marfan syndrome using the revised Ghent criteria
☐ Talipes equinovarus
☐ Hypertelorism
☐ Craniosynostosis
☐ Cleft palate
☐ Bifid uvula
☐ Blue sclerae
☐ Arterial tortuosity
☐ Patent ductus arteriosus
☐ Velvety/translucent skin
☐ Easy bruising
☐ Widened atrophic scars
☐ Spontaneous organ rupture
☐ Aortic Dimensions mm, Z-score
☐ Fractures; describe:
☐ Hearing loss
☐ Stroke
☐ Other aneurysm; describe:
☐ Other:

Connective Tissue/Cerebrovascular Disease Genetic Testing Patient Information (continued)

Patient Name (Last, First Middle)	Birth Date (mm-dd-yyyy)

Clinical History (continued)

Ghent Systemic Score Calculation for Marfan Syndrome

Feature	Value	Enter Value if Present	Feature	Value	Enter Value if Present
Wrist **and** thumb sign	3		Scoliosis or thoracolumbar kyphosis	1	
Wrist **or** thumb sign	1		Reduced elbow extension	1	
Pectus carinatum	2		3 of 5 facial features: • dolichocephaly • enophthalmos • downslanting palpebral fissures • malar hypoplasia • retrognathia	1	
Pectus excavatum or chest asymmetry	1				
Hindfoot valgus **and** flat foot (pes planus)	2				
Isolated pes planus (flat foot)	1				
Pneumothorax	2				
Dural ectasia	2		Skin striae	1	
Protrusio acetabulae	2		Myopia > 3 diopters	1	
Reduced upper/lower segment **and** increased armspan/height	1		Mitral valve prolapse	1	
Total Value: 0.00					

List any additional features present:

Is this a postmortem specimen?
☐ Yes ☐ No If "Yes," attach autopsy report if available.

Biological Family History

Are there similarly affected biological relatives? ☐ Yes ☐ No
If "Yes," indicate relationship and symptoms:

Have any biological family member had genetic testing? ☐ Yes*** ☐ No ☐ Unknown
***FMTT / Familial Variant, Targeted Testing should be ordered when there is a previous positive genetic test result in the biological family. Contact the lab for ordering assistance.**

History of consanguinity
☐ No ☐ Unknown ☐ Choose not to disclose ☐ Yes; relationship details:

Patient Ancestry

Ancestry: ☐ Unknown ☐ Choose not to disclose

New York State Patients: Informed Consent for Genetic Testing is required. See Informed Consent for Genetic Testing (T576) or Informed Consent for Genetic Testing – Spanish (T826).

Print